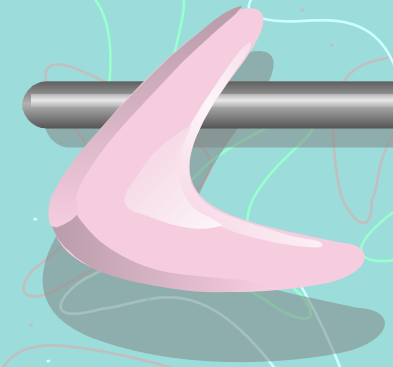
Genetic Screening for Cystic Fibrosis



A New Choice for You and Your pregnancy

Goals for this Discussion

- Present information about a new screening test for a common genetic disease
- Discuss the nature of this disease, cystic fibrosis
- Discuss how testing can be accomplished
- Discuss what the test will tell you
- Discuss what the test won’t tell you
- Discuss the risks of genetic testing
- Discuss how the test results can be managed

What is Cystic Fibrosis (CF)?

- Cystic fibrosis is an inherited disease which may lead to the development of very thick, easily dried out mucous in nose, lungs, and intestines
- Disease may be mild or very severe
  - Generally it is a chronic life-long illness requiring some kind of lifetime treatment
  - No present cure

Areas affected by CF

- Lungs
  - Thick clogging mucous
  - Frequent coughing and wheezing
  - Frequent pneumonia and bronchitis
- Chronic sinus infections
- Digestion problems
- Altered reproduction
  - Men may be infertile; pregnancy may be risky for affected women

Healthcare for CF patients

- Daily breathing treatments, chest therapy, and medications
- Daily vitamins, enzymes for digestion, and careful diet
- Treatment or preventative treatment for infections
- Treatment for development of diabetes

Outcome of Effective Treatment for CF

- Normal or near normal growth
- Normal intellectual development
- Increasing lifespan
  - About 50% of people with CF live to age 30
  - Babies born now may have average length of life to over 50

Diagnosing CF

- About 1 in 2500 to 1 in 3000 Caucasian babies have CF
- In a child, the diagnosis is made by measuring the amount of salt in the sweat
- Also made by the clinical appearance of the condition
- DNA testing may determine the genetic cause of the disease

What causes CF?

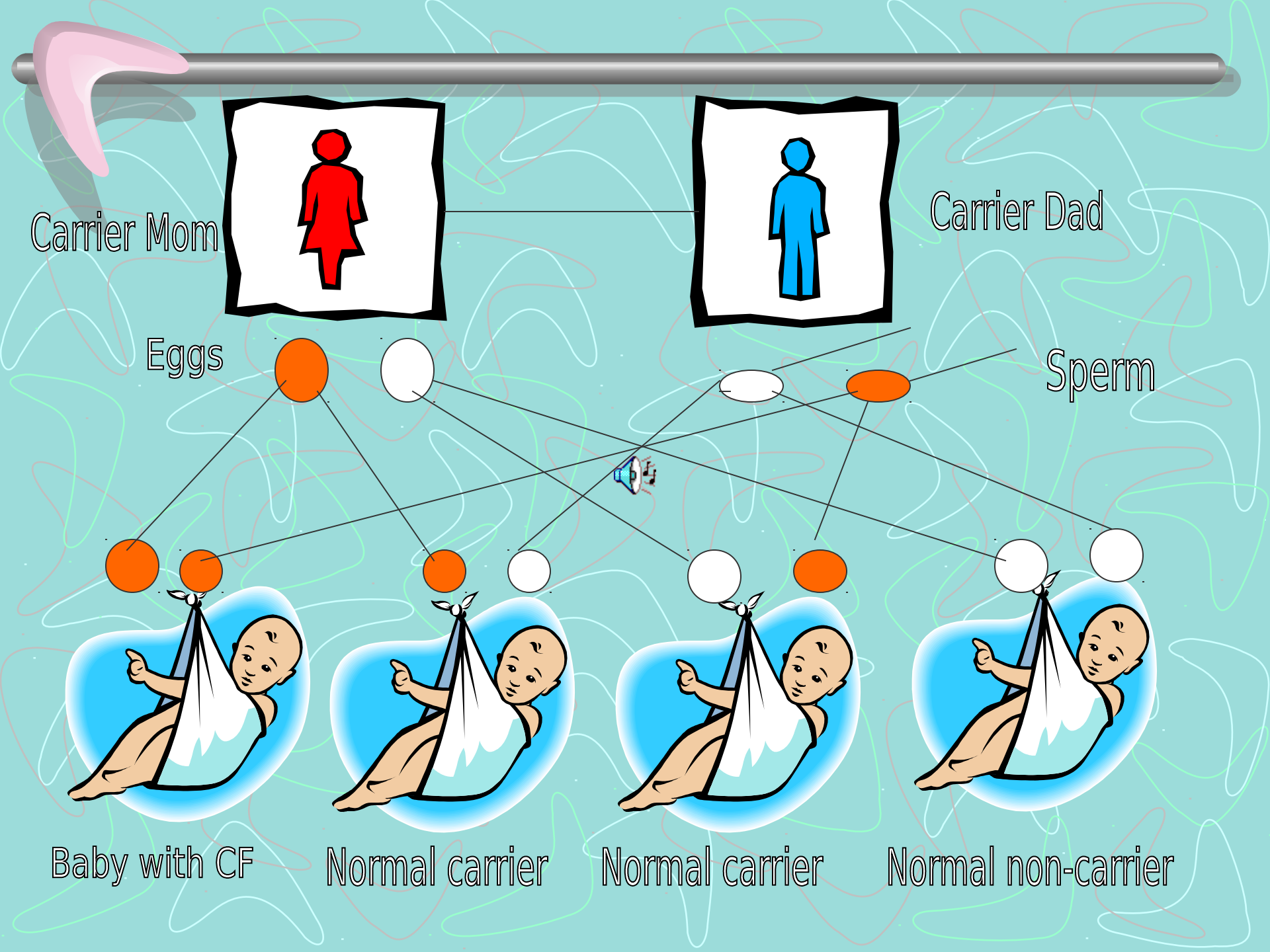
- CF is an inherited (genetic) condition causes by a **pair** of genes which are not working properly
  - Genes are genetic material passed from parents to children
  - Genes determine how and what proteins are made in the body
  - Genes are made up of DNA
  - Changes in DNA which change the protein the gene makes are called **mutations**

Genetics of CF

- Genes are inherited in pairs, one from the mother, one from the father
  - If the parents carry one mutated gene and one normal gene, they are called carriers
    - **Carriers of one mutated gene are completely normal**

Genetics of CF

- Cystic fibrosis occurs when **both** genes for the CF protein are changed (mutated)
  - **Only if the baby gets an abnormal gene from mother AND father will it have cystic fibrosis**


Carrier Mom
Carrier Dad
Eggs
Sperm
Baby with CF
Normal carrier
Normal carrier
Normal non-carrier

What is your chance of being a CF carrier?

- The chance for a person to be a carrier of CF depends largely on their ethnic background
  - Highest carrier rates in people of Caucasian and Ashkenazi Jewish background
    - Chance is 1 in 29 that people in those groups carry CF
    - Includes people whose background is from England, Scotland, Wales, Scandinavia, Europe

Chance of Being a CF Carrier by Ethnic Background

Ethnic group	Affected child	Carrier rate	**Ability to Detect mutation**
Europ. Cauc	1/3000	1/29	80%
Ash. Jewish	1/3000	1/29	97%
Hispanic Am	1/9200	1/46	57%
African Am	1/15,000	1/65	69%

Other Effects of Ethnic Background

- The likelihood that testing can detect a mutation depends on the ethnic group
  - Non-Caucasian or non-Jewish groups **<u>are already at low risk for having children with CF</u>**
  - Testing may not add much additional information about these groups

CF Carrier Screening

- Testing is available to provide information about your risk for being a CF carrier
- This is termed “screening” testing because it is testing people who do not have the condition

What if there is a family history of CF?

- You and your husband will need genetic counseling about the family history
- Genetic testing for CF will be offered if desired
- You do not fall into the general screening program for CF because you are already a higher risk

Is there a benefit to me to have Genetic Screening for CF?

- Genetic screening can identify if you are at higher risk than you thought for having a baby with cystic fibrosis
- Genetic screening may be reassuring to you that you have a lower chance of having a baby with this serious problem

How do I find out if I am a carrier for CF?

- You must sign a consent form stating that you understand what testing means and that you want to be tested
- A blood specimen is needed
  - May be part of New Mothers' labs if not yet drawn
  - If those labs already drawn, another tube must be drawn
- If you need to think about testing and want to wait, fresh blood may need to be drawn later

Genetic Testing Process

- IF the mother gives consent (checks “Yes” on the consent form), then the blood is tested
  - DNA testing is done on the blood
  - Results come back in approximately 3 weeks
- IF the mother does **NOT** give consent (checks “No” on the consent form), then **no** blood is drawn and **no** testing for CF is done

The results show: a mutation is not found

- If no mutation is found, the risk is reduced and no further testing is done
  - **No further CF testing is needed in any other pregnancy** unless the father changes and has a family history of CF

What does it really mean: No mutation?

- No mutation means that no abnormality was found when your DNA was studied to see if it showed multiple different mutations
- **HOWEVER**, testing cannot say 100% you are not a carrier; it just reduces your chance of being a carrier and having an affected baby
  - A woman of European Caucasian background without family history of CF is tested for CF. No mutations are found
    - Her risk of being a carrier changes from 1 in 29 to 1 in 140 with a very low chance for an affected baby

The results show: a mutation **is** found

- If the test finds a mutation (it is a positive test), then **you are a carrier** of a specific known change in the DNA for CF
- The next step is to find out if your partner is also a carrier
- **Only if both parents are carriers can you have a baby with CF**

What if my partner is also a carrier?

- If both parents are found to be carriers of mutations in CF, then they need further information
  - Detailed genetic counseling about their 1 in 4 risk of having an affected baby
  - Prenatal diagnosis including amniocentesis or other testing to identify if the baby is affected may be of interest to some couples
  - The baby can also have genetic testing at birth

Benefits of Prenatal Diagnosis for CF

- Family can prepare for the birth of a baby with special needs
- Baby will benefit from early treatment from birth to improve health
- Some couples may not wish to continue a pregnancy if they know their baby is affected with cystic fibrosis

New advances in CF

- Even if mutations are found in both parents, it does not always predict how severely affected a child will be
  - Medical advances have improved the lifetime care for CF patients and will continue to do so

What if my baby's father is unavailable or unknown?

- If the father is not available because he is stationed somewhere else, blood can be drawn from him and mailed back for testing
- If the father is unknown, the risk to the baby is based only on the mother's carrier risk
  - It is possible that an affected baby may be missed if the father is not tested

Should I get Genetic screening for CF?

- The choice is up to you!
  - The greatest value is if you belong to an ethnic group which has a higher risk of having CF
    - Includes European Caucasian and Ashkenazi Jewish
  - Other ethnic groups have such a low risk of CF that testing may add very little to their knowledge
    - Includes African Americans, Hispanic Americans, and Asian Americans

Do I **have to have** Genetic Screening for CF?

- No!
- If you do not want genetic screening for CF, mark “NO” on the consent form
  - **Your blood will not be tested unless you consent**
- If you decide later you want testing, you can give your consent and a new specimen of blood will be drawn
- If you don’t want to be tested, you will still get the same good prenatal care that you would get if you did get tested
  - There’s no penalty for not testing!

Reminder about testing

- All genetic tests are specific—they only look for one particular condition
  - If you do not have a mutation for CF, it does not mean that you could not have a mutation for some other genetic condition entirely
- There is no testing available for all genetic disorders
- If you have a family history of some other genetic problem, you may benefit from genetic counseling to discuss what is your risk of being a carrier of that problem